

Variant: NM_001130987.2(DYSF):c.3114G>A (p.Pro1038=)

Version: 1.0

CA222150 [↗](#)

94299 (ClinVar) [↗](#)

Gene: DYSF ([HGNC:8291](#))

Condition: autosomal recessive limb-girdle muscular dystrophy
([MONDO:0015152](#))

Inheritance Mode: Autosomal recessive inheritance

UUID: 9e5bf7d2-f973-4ada-b086-7cb8c9e3f36a

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HGVS expressions

NM_001130987.2:c.3114G>A

NM_001130987.2(DYSF):c.3114G>A (p.Pro1038=)

NC_000002.12:g.71570627G>A

CM000664.2:g.71570627G>A

NC_000002.11:g.71797757G>A

CM000664.1:g.71797757G>A

NC_000002.10:g.71651265G>A

NG_008694.1:g.122005G>A

ENST00000698057.1:c.486G>A

ENST00000258104.8:c.3060G>A

ENST00000410020.8:c.3114G>A

ENST00000258104.7:c.3060G>A

ENST00000394120.6:c.3063G>A

ENST00000409366.5:c.3063G>A

ENST00000409582.7:c.3111G>A

ENST00000409651.5:c.3156G>A

ENST00000409744.5:c.3021G>A

ENST00000409762.5:c.3111G>A

ENST00000410020.7:c.3114G>A

ENST00000410041.1:c.3114G>A

ENST00000413539.6:c.3153G>A

ENST00000429174.6:c.3060G>A

ENST00000461565.1:n.226G>A

NM_001130455.1:c.3063G>A

NM_001130976.1:c.3018G>A

NM_001130977.1:c.3018G>A

NM_001130978.1:c.3060G>A

NM_001130979.1:c.3153G>A

NM_001130980.1:c.3111G>A

NM_001130981.1:c.3111G>A

NM_001130982.1:c.3156G>A

NM_001130983.1:c.3063G>A

NM_001130984.1:c.3021G>A

NM_001130985.1:c.3114G>A

NM_001130986.1:c.3021G>A

NM_001130987.1:c.3114G>A

NM_003494.3:c.3060G>A

NM_001130455.2:c.3063G>A
NM_001130976.2:c.3018G>A
NM_001130977.2:c.3018G>A
NM_001130978.2:c.3060G>A
NM_001130979.2:c.3153G>A
NM_001130980.2:c.3111G>A
NM_001130981.2:c.3111G>A
NM_001130982.2:c.3156G>A
NM_001130983.2:c.3063G>A
NM_001130984.2:c.3021G>A
NM_001130985.2:c.3114G>A
NM_001130986.2:c.3021G>A
NM_003494.4:c.3060G>A

Likely Benign

Met criteria codes 1

BS1

Not Met criteria codes 3

PM3

BP4

BP7

Evidence Links 0

Expert Panel

Limb Girdle Muscular Dystrophy VCEP

Criteria Specification Information

Criteria Specification: ClinGen Limb Girdle Muscular Dystrophy Expert Panel Specifications to the ACMG/AMP Variant Interpretation Guidelines for DYSF Version 2.0.0

Criteria Specification Approval History

Criteria Specifications for this VCEP

Evidence submitted by expert panel

Limb Girdle Muscular Dystrophy VCEP

The NM_003494.4: c.3060G>A p.(Pro1020=) variant in DYSF, which is also known as NM_001130987.2: c.3114G>A p.(Pro1038=), is a synonymous (silent) variant that is not located in a splice region (outside of the first and the last 3 bases of the exon). This variant was detected in a heterozygous state in one individual with suspected LGMD and disease range dysferlin expression. This individual also had two additional variants in DYSF in unknown phase (PM3 and BP2 not met; PMID: 30564623, LOVD Individual #00222264; Jain Foundation Dysferlin Registry internal data communication). The filtering allele frequency for this variant is 0.001555 in gnomAD v4.1.0 (the lower threshold of the 95% CI of 162/91072 South Asian chromosomes), which is higher than the VCEP threshold of 0.001 (BS1). The SpliceAI prediction score for this variant is 0.17, which is greater than the VCEP threshold of <0.05 (BP4 not met). In summary, this variant meets the criteria to be classified as Likely Benign for autosomal recessive limb girdle muscular dystrophy based on the ACMG/AMP criteria applied, as specified by the ClinGen LGMD VCEP (specifications v2.0.0; 01/23/2026): BS1.

Met criteria codes

BS1



The filtering allele frequency for this variant is 0.001555 in gnomAD v4.1.0 (the lower threshold of the 95% CI of 162/91072 South Asian chromosomes), which is higher than the VCEP threshold of 0.001 (BS1).

Not Met criteria codes

PM3



This variant was detected in a heterozygous state in one individual with suspected LGMD and disease range dysferlin expression. This individual also had two additional variants in DYSF in unknown phase (PM3 and BP2 not met; PMID: 30564623, LOVD Individual #00222264; Jain Foundation Dysferlin Registry internal data communication).

BP4			The SpliceAI prediction score for this variant is 0.17, which is greater than the VCEP threshold of <0.05 (BP4 not met).
BP7			The SpliceAI prediction score for this variant is 0.17, which is greater than the VCEP threshold of <0.05 (BP4 not met). Per VCEP, BP4 needs to be met to apply BP7.

Curation History



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